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The policy of public health genomics in Italy

Benedetto Simone[a], Walter Mazzucco[a,b], Maria Rosaria Gualano[n], Antonella Agodi[c], Domenico Coviello[d], Francesca Dagna Bricarelli[e], Bruno Dallapiccola[f], Emilio Di Maria[g,h], Antonio Federici[i], Maurizio Genuardi[j,k], Liliana Varesco[l], Walter Ricciardi[a], Stefania Boccia[a,m,*], for the GENISAP Network[1]

[a] *Institute of Hygiene, Università Cattolica del Sacro Cuore, Largo F. Vito 1, 00168 Rome, Italy*
[b] *Department of Health Promotion Sciences, University of Palermo, Piazza Marina 61, 90133 Palermo, Italy*
[c] *Dipartimento di Anatomia, Biologia e Genetica, Medicina Legale, Neuroscienze, Patologia Diagnostica, Igiene e Sanità Pubblica, "Gian Filippo Ingrassia", Università degli Studi di Catania, Via S. Sofia 87, Comparto 10 Edificio C, 95123 Catania, Italy*
[d] *S.C. Laboratorio di Genetica Umana, E.O. Ospedali Galliera di Genova, Mura delle Cappuccine 14, 16128 Genoa, Italy*
[e] *Dipartimento Ligure di Genetica, E.O. Ospedali Galliera, Via Volta 6, 16128 Genoa, Italy*
[f] *Ospedale Bambino Gesù, IRCCS, Piazza Sant'Onofrio 4, 00165 Rome, Italy*
[g] *Department of Health Sciences, University of Genova, Via A. Pastore 1, 16132 Genoa, Italy*
[h] *Division of Medical Genetics, E. O. Ospedali Galliera di Genova, Mura delle Cappuccine, 14, 16128 Genova, Italy*
[i] *Ministero della Salute, Lungotevere Ripa 1, 00153 Rome, Italy*
[j] *Unit of Medical Genetics, Department of Clinical Pathophysiology, University of Florence, Viale G. Pieraccini 6, 50139 Florence, Italy*
[k] *FiorGen Foundation for Pharmacogenomics, Via Luigi Sacconi 6, 50019 Sesto Fiorentino, Florence, Italy*
[l] *Centro Tumori Ereditari, IRCCS AOU San Martino – IST, Largo R. Benzi 10, 16123 Genoa, Italy*
[m] *IRCCS S. Raffaele Pisana, Via della Pisana 235, 00163 Rome, Italy*
[n] *Department of Public Health Sciences, University of Turin, via Santena 5/bis, 00126 Turin, Italy*

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ABSTRACT

Italy has a monitoring system for genetic testing, consisting in a periodic census of clinical and laboratory activities performed in the country. The experience is limited, however, concerning the translation of genomic testing for complex diseases into clinical practice. For the first time the Italian Ministry of Health has introduced a policy strategic plan on genomics and predictive medicine within the 2010–2012 National Prevention Plan. This achievement was supported by the Italian Network for Public Health Genomics (GENISAP) and will likely contribute to the integration of public health genomics into health care in the country. Our experience might be of interest not only in Italy, but in other high-income countries, struggling to keep a healthy economy and healthy citizens.

* Corresponding author at: Institute of Hygiene, Università Cattolica del Sacro Cuore, L.go F. Vito 1, 00168 Rome, Italy. Tel.: +39 06 35001527; fax: +39 06 35001522.

E-mail addresses: benedetto.simone@yahoo.it (B. Simone), walter.mazzucco@unipa.it (W. Mazzucco), mariarosaria.gualano@unito.it (M.R. Gualano), agodia@unict.it (A. Agodi), coviello@unige.it (D. Coviello), francesca.dagna@galliera.it (B. Dallapiccola), emilio.dimaria@unige.it (E. Di Maria), an.federici@sanita.it (A. Federici), m.genuardi@dfc.unifi.it (M. Genuardi), liliana.varesco@istge.it (L. Varesco), direzioneigiene@rm.unicatt.it (W. Ricciardi), sboccia@rm.unicatt.it (S. Boccia).

[1] List of members of the GENISAP Network can be found on http://istituti.unicatt.it/igiene_1830.html.

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1. Background

Italy has a population of 60.6 million. It has been ranked the world's twenty-fourth most-developed country in 2010 [1], and its quality of life index, gross domestic product (GDP), nominal GDP per capita and standard of living are among the highest in the world [2–4]. Once a country of mass emigration, Italy is today home to over 4 million foreign national residents registered with authorities, 7.1% of the country's population [1].

1.1. Health system

Since 1978 Italy has had a public healthcare system based on principles of universalism, comprehensiveness and solidarity in funding. In 2008 healthcare expenditure amounted to 9.5% of the national GDP; the health system is highly evolved and ranks 2nd worldwide, with the 3rd best healthcare performance [1]. Most of the public health care current expenditure is still dedicated to the hospital sector, which accounts for approximately 46% of the overall healthcare budget. Although some progress has been made in the past years, prevention only absorbs 4% of the overall healthcare expenditure [1,5].

Healthcare today is provided by a mixed public-private system and is administered on a regional basis. The devolution of powers and competences at a sub-national level, which was started with the reform of "Title V" of the Italian Constitution in 2001, required the establishment of solid links between national and regional institutions. The role of central government is conceived as to set the so-called "essential levels of care" (*Livelli Essenziali di Assistenza*, LEAs), which must be guaranteed to all residents [6].

2. Current scenario on genetic and genomic testing in Italy

2.1. Clinical and prenatal genetics

To our knowledge, Italy is the only country in Europe and one of the very few in the world where a monitoring system for the use of genetic tests was implemented since the mid-80s. The Italian Society of Human Genetics (SIGU, *Società Italiana di Genetica Umana*) has been carrying out a periodic census of clinical and laboratory activities performed in the country. The last census, in 2007, covered 217 molecular genetic laboratories, 171 cytogenetic laboratories and 102 clinical genetic services throughout the country (total: 490). Data were collected from 278 responding centres (respondence rate: 57%). Only 28% of the responding centres were certified according to quality standards. About 560,000 genetic tests, including 311,069 cytogenetic and 248,691 molecular analyses of 556 genes, were recorded. The foetal karyotype was examined on either chorionic villi or amniocytes in about one of every 4.4 at term pregnancies. Only the 11.5% of cytogenetic analyses and the 13.5% of molecular tests were accompanied by genetic counselling. Low congruity was found between clinical diagnoses and laboratory results, suggesting that the request of genetic tests may not be appropriate in several instances [7].

2.2. Complex diseases

Predictive genetics have currently few applications in clinical practice. Predictive tests have raised some interest in public health only in the case of high-penetrance genetic variants associated with common types of cancer (breast/ovarian and colorectal cancer syndromes) and, to a lesser extent, the maturity onset diabetes of the young (MODY).

Testing for low-penetrance polymorphisms is still fragmentary, and a structured and organic experience in Italy is still missing.

The Institute of Hygiene of Università Cattolica in Rome has partnered the Public Health Genomics European Network (PHGEN) in 2006 and founded in 2007 the Italian Network for Public Health Genetics, later named GENISAP Network, with the objectives of generating knowledge, monitoring the predictive genetic testing activities in Italy, assessing their appropriateness, performing cost-effectiveness analyses and contributing to the development of evidence-based recommendations and guidelines on the translation of genomic technologies in clinical settings [8,9]. The GENISAP Network, four years later, counts around 50 experts from different backgrounds.

Recent results produced by some of the members of the GENISAP Network have shown that molecular laboratories testing for hereditary breast/ovarian and colorectal cancers in four Italian Regions (Abruzzi, Liguria, Tuscany, Latium) are not coordinated, as there are no regional (and national) guidelines or plans for the delivery of these tests, and quality is not monitored in a systematic way. Furthermore, there is a very wide inter-laboratory heterogeneity in terms of procedures, costs and turnaround times. Lastly, genetic tests for well-established hereditary syndromes (namely, *BRCA*-related breast/ovarian cancers and Lynch syndrome) appear markedly under-prescribed compared to population estimates of their incidence [10].

A more systematic approach to care of women at risk of breast cancer has recently been undertaken by Emilia-Romagna Region which has organised a network, based on a "hub and spoke" model, involving genetic and senology units in integrated activities [11]. This model includes quality monitoring of clinical variables (*e.g.* referral appropriateness and efficacy of clinical pathways) and establishes genetic testing management strategies.

Another study promoted by the GENISAP Network focused on the use of genetic tests for the assessment of the risk of developing thromboembolic events [methylenetetrahydrofolate reductase (*MTHFR*) C677T polymorphism, Factor V Leiden, prothrombin G20210A]. The conclusion was that these tests are highly prescribed, although scientific evidence does not support their use for assessing the individual thromboembolic risk [12]. This is particularly true with regard to the *MTHFR* gene: although current literature is concordant that *MTHFR* polymorphic variants do not affect significantly the risk of thrombosis, it is prescribed within the panel of thrombophilia genes, which is by large the most frequently prescribed susceptibility test in Italy [7].